Study of alkaptonuria in residences of Divlag village

Abstract

Background and Objective: Alkaptonuria is a lifelong disease with no known treatment. Since some cases of this rare disease were diagnosed in Divlag village and due to autosomal recessive hereditation of the disease, social factors such as marriage with relatives may play role in this issue. Also, there are no concessive information on this issue. Thus, Divlag village was selected and assessed.

Methods: All Divlag village population referring to health house or village health center were invited to this study. 600 of 1100 total population participated in this study. Urine sample were taken from all participants and positive cases for Alkaptonuria were diagnosed by urine analysis.

Results: Mean age of participants was 31.34 ± 19.2 years old (50.7% male and 49.3% female). 11 cases (1.8%) reported Alkaptonuria in their first degree relatives. Marriage with relatives had a 28.7% prevalence. 3 Alkaptonuria cases (0.5%) were diagnosed including 2 male (24 and 49 years old) and a female (63 years old). Clinical manifestations included neck pain (in 2 cases), neck limited range of motion (in 2 cases), lumbar vertebral column pain (in 2 cases), back pain (in 2 cases), reduction of lumbar lordosis (in 2 cases), knee pain (in 2 cases), morning stiffness (in 2 cases), and reduction of cervical lordosis (in 1 case). Observed pigmentation in the 49 and 63 years-old patients were as follows: in both ears and between first and second finger of both hands in both patients, in one sclera of 49 years-old patient and in nasal rim and on checks of second patient (mild pigmentation) in 63 years-old patient. In the 24 years-old patient, suspicious pigmentation of ears were observed. Urine of all three patients turned to black when exposed to air and sun light.

Conclusion: Three new Alkaptonuria cases were diagnosed in this study. Further assessments are suggested for further evaluation of prevalence and incidence of Alkaptonuria in Ardabil as well as genetic study of these patients.

Key Words: Alkaptonuria, Homogentisic acid.